4 Dimensional Phenotyping

Gareth Baynam[1,2], Mark Walters[3], Peter Claes[4], Stephanie Kung[2], Peter LeSouef[2], Hugh Dawkins[5,6,7], Matthew Bellgard[8], Marta Girdea[9], Peter Robinson[10], Andreas Zankl[11], David Gillett[3], Jack Goldblatt[1,2]

[1]Genetic Services of Western Australia, Princess Margaret & King Edward Memorial Hospitals, Perth, Western Australia, Australia, [2]School of Paediatrics and Child Health, University of Western Australia, Perth, Western Australia, Australia [3]Cranio-Maxillo-Facial Unit, Princess Margaret Hospital for Children, Perth, Western Australia, Australia, [4]P. Claes, K.U. Leuven, Medical Imaging Research Centre, Faculty of Engineering, Leuven, Belgium, [5]Office of Population Health Genomics, Department of Health, Perth, Western Australia, Australia, [6]Centre for Population Health Research, Curtin Health Innovation Research Institute, Curtin University of Technology, Perth, Western Australia, Australia, [7]School of Pathology and Laboratory Medicine, University of Western Australia, Perth, Western Australia, Australia, [8]Center for Comparative Genomics, Murdoch University; Department of Computer Science, University of Toronto, Ontario, Canada, [9]Institut für Medizinische Genetik und Humangenetik, Charité – Universitätsmedizin, Berlin, Germany, [10]The University of Queensland, UQ Centre for Clinical Research, Queensland, Australia

Analysis of the clinical phenotype: history, examination and investigation, is the physician's daily work[1]. Documentation of patient phenotypes has become particularly to clarify genotype–phenotype relationships that are increasingly being investigated by the use of high throughput molecular technologies. To improve the clinical utility of emerging -omics technologies, such as next generation sequencing, there is a parallel and increasing need for efficient, and preferably objective and standardised, documentation of the clinical phenotype. This will require time-efficient tools for ascertaining phenotypic information, incorporating this information into clinical work flows, and for integrating with -omics analyses. This presentation will focus on aspects of phenotyping that are of particular relevance to rare diseases and which have implications for common diseases. Firstly, for illustrative purposes, it will discuss 3-dimensional facial analysis: a fine scale, objective, scalable, portable, cost efficient, non-invasive and non-irradiating phenotypic assessment[2]. Secondly, it will describe the "4th dimension" of bed-side capture of standardized phenotypic information, workflow integration and emerging decision support tools.

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